

Speaker: Jannine Cody, Ph.D.

Topic: Birth Defect Support Groups: What Do They Do and How Do They Help Families? (Genetic Support Groups, Myths, Realities and Rewards)

Objectives:

- 1) Name three specific services or benefits that birth defect support groups can provide to families.
- 2) List three reasons that support groups vary in their emphasis.
- 3) List two methods for locating a support information about a specific support group.

Outline:

- 1) Functions of a support group.
 - a) Community
 - b) Education
 - c) Research
- 2) Reasons for variability among support groups.
- 3) Why does a family join a support group?
- 4) How support groups can improve medical care.
- 5) How to find a support group.
- 6) How to start a support group

Abstract: Support groups exist for a multitude of genetic conditions. The focus and mission of different support groups varies, but most perform three basic functions. They provide a sense of community for affected individuals and their families. They educate families, the public and policy makers about life with their respective conditions. Support groups also support research by either organizing potential participants or by directly funding research. The degree to which any one group performs these functions is dependant upon several factors, the most important of which is the nature of the condition itself. The age and size of the group greatly impacts the programs and emphases within the group.

Often when families are first given a diagnosis, they have no idea that a support group might exist to help them understand and live with the condition. It is often up to the medical professionals who know that support groups exist and know how to find them to first inform a family. Although no two genetic support groups are the same, each has its place in the lives of its members.

Resources for Genetic Support Groups

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Alliance of Genetic Support Groups

4301 Connecticut Ave. NW, Suite 404

Washington, DC 20008

1-800-336-GENE, (202) 966-5557, Fax (202) 966-8553

<http://www.geneticalliance.org>

NORD (National Organization for Rare Disorders)

P.O. Box 8923

New Fairfield, CT 06812-8923

1-800-999-NORD, fax 203-746-6481

<http://www.NORD-rdb.com/~orphan>

Rare Genetic Diseases In Children: An Internet Resource Gateway

Paul Murphy, pmurf@ix.netcom.com

<http://mcr4.med.nyu.edu/~murph01/homenew.htm>

NIH Office of Rare Diseases

<http://rarediseases.info.nih.gov/ord/pages>

Where can I go to Find Disability-Related Information?@

<http://www.icdi.wvu.edu/Others.html>

Starting & Sustaining Genetic Support Groups by Joan O. Weiss and Jayne S. Mackta, Johns Hopkins University Press, Baltimore, MD, 1996

Speaker: Maricela Aguilar, R.N., M.S.N.

Topic: Cultural Barriers to Receiving Genetic Services

Objectives:

- 1) Describe Cultural Competency as it relates to the provision of health care.
- 2) Discuss self-awareness as it relates to the practice of genetics.
- 3) Describe Mexican-American cultural beliefs and health practices that affect the utilization of health services.
- 4) Identify cultural and non-cultural issues affecting access and utilization of genetic services and other health care among Mexican Americans in South Texas.

Outline:

- 1) Cultural Competency
- 2) Self-awareness and the Genetic Service Provider
- 3) Mexican American Cultural Beliefs and Traditional Healing Practices
in South Texas
 - a) Background on Ethnographic Study
 - b) Who Are the Mexican Americans?
 - c) Cultural Influences and Genetic Service Utilization
 - d) Curanderismo
- 4) Barriers to Genetic Services in South Texas
- 5) Bridging the Gap: Reducing Barriers to Genetic Services

Abstract: Participants in this session will explore individual and systems issues that affect cultural competency and methods to evaluate self-awareness of cultural competency in ten primary areas. Cultural beliefs and traditional healing practices among Mexican Americans in South Texas, specific to birth defects, will be presented. This information is based on a recent ethnographic study of Medicaid genetic service clients, genetic service providers and traditional healers in South Texas and Mexico. The goal of the study, funded by the U.S. Maternal and Child Health Bureau, was to reduce barriers that affect access and utilization of genetic services in South Texas. The barriers identified in the study and recommendations for reducing them will also be discussed.

Speaker: John Teichgraeber, M.D.

Topic: Local, National, and International Program for the Repair of Cleft Lip/Palate

Objectives:

- 1) Identify three challenges unique to families in developing countries whose children are affected by facial deformities.
- 2) Describe the services offered by agencies performing medical missions in the repair of craniofacial birth defects.
- 3) Name five ways in which corrective surgery improves quality of life or health for children with related defects.

Abstract:

The presentation will review the Austin Smiles experience in Central America. The mission has been ongoing for a decade, with its primary focus in El Salvador. The organization of the mission is unique in that Austin Smile's visits El Salvador three times a year and returns to the same hospital each time. They have operated on over 1000 patients. The set-up of the program is such that it is designed to provide continuity of care. Moreover comprehensive care of the children with cleft deformities is an out growth of the missions design. The mission is also closely coordinated with the Military Hospital in San Salvador and the Minister of Infant and Maternal Services who provide the local facilities and services.

Speaker: Jacqueline Hecht, Ph.D.

Topic: Genetic Counseling

Objectives:

- 1) Describe categories for which genetic counseling is sought.
- 2) Describe the genetic counseling process.
- 3) Describe the grief reaction.
- 4) Define successful genetic counseling outcomes.

Outline:

- 1) Definition of genetic counseling
- 2) The goals are to help the individuals and or families to:
 - a) Comprehend the medical facts.
 - b) Understand the risk of recurrence.
 - c) Understand the options for dealing with the recurrence risk.
 - d) Choose an appropriate course of action and act accordingly.
 - e) Make the best Possible adjustment
- 3) The Genetic Counseling process includes:
 - a) Accurate diagnosis
 - b) Clear communication
 - c) Nondirective counseling
 - d) Respect for and adjustment to the family's psychological and emotional turmoil
 - e) The family unit as a focus of health care
 - f) Extended family counseling
 - g) Positive contributions to patient management
 - h) Genetic counseling is not a circumscribed, isolated clinical effort
 - i) Prognostic and treatment options
 - j) Team approach.
- 4) Impact/Burden of Genetic Disorders
 - a) Grief Reaction
 - i) Shock
 - ii) Anxiety
 - iii) Guilt/Anger/Shame/Embarrassment
 - iv) Adaptation/Re-organization.
 - b) Indications for Genetic Counseling.

- i) Mendelian traits
 - ii) Chromosomal disorders
 - iii) Polygenetic disorders
 - iv) Common congenital malformations
 - v) Familial disorders with unknown modes of inheritance
 - vi) Syndromes of unknown etiology
 - vii) Undiagnosed familial disorders
 - viii) High risk relatives.
 - ix) Premarital counseling for familial disease in intended spouse
 - x) Advanced maternal age pregnancies.
 - xi) Exposure to possible teratogens or mutagens
 - xii) Consanguineous matings
 - xiii) Adoption
 - xiv) Paternity determination
 - xv) Screening or artificial insemination donors
 - xvi) Prospective counseling
- 5) To identify the indications for genetic referral.
 - 6) To learn the genetic counseling process.
 - 7) To learn the psychosocial issue related to genetic counseling.
 - 8) To learn successful genetic counseling outcomes.

Speaker: Hallie Duke, Ph.D.

Topic: "On the Right Track"-Family Centered Planning"

Objectives:

- 1) Define Family-Centered Planning and team planning in the context of school planning.
 - a) What is Family-Centered Planning?
 - b) What does Family-Centered Planning mean for a family?
 - c) How does the Family-Centered approach benefit health care providers?
 - d) What is a Team Planning Process?
- 2) Describe the AOn the Right Track@plan for a pilot study.
 - a) The primary goal of the research:
 - b) Pilot Project Elements
 - c) Basic Steps in Pilot
- 3) Name two ways team planning can prevention of secondary conditions for children with special health care needs.
 - a) How family-centered planning prevents secondary conditions
 - b) What we hope to achieve.
 - c) Where we see it going.

Abstract: AOn the Right Track@focuses on communication and planning to prevent secondary conditions and improve quality of life for people with disabilities. The two intervention strategies are 1) family-centered collaborative planning and 2) the use of a planning tool to enhance information sharing between families, health care providers, and educators. In the AOn the Right Track@family-centered, collaborative planning method, parents, teachers, and medical service providers (e.g., therapists, physicians, etc.) will be brought together as a team with a facilitator to establish mutual goals for the child and to develop action plans for the achievement of those goals. The facilitator will act as an advocate for effective planning, facilitating communication among team members to ensure a balance of information sharing. This method of planning takes into account the family's needs, priorities, and capabilities. The on-line planning software that was developed for this project has built-in features that reflect the collaborative and family-centered themes central to this project.

The presentation will provide information about and discussions of the primary concepts of the family-centered approach to the AOn the Right Track@research pilot as well as detailed information about the implementation of the intervention.